April 7 & 8

Stoney Creek Inn and
Conference Center
Johnston, IA

What do YOU think?

A COMMUNITY DISCUSSION OF THE IOWA NEWBORN SCREENING PROGRAM

IDPH
Iowa Department of Public Health

Pre-Event Booklet

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Dear Reader,

The information in this book is based on recent research, expert opinion, and parent testimony. We are sharing this information so that you can reflect and prepare for the topics that will be discussed during the Community Engagement Event.

There many topics covered in this booklet. However, it's not possible to explore every issue in depth within our page limits. Please feel free to explore any of the other information on our website [https://idph.iowa.gov/newborn-screening] and on the National Newborn Screening website [http://www.babysfirsttest.org/].

Thank you for taking the time to read this booklet and think about this information. Please contact us with questions.

Sincerely,
the Iowa Newborn Screening Program

Iowa Department of Public Health
Protecting and Improving the Health of Iowans

Gerd W. Clabaugh, MPA
Director

Kim Reynolds
Governor

Adam Gregg
Lt. Governor

February 28, 2018

Dear Community Engagement Event Participants,

Public health is a partnership of local public health, the Iowa Department of Public Health (IDPH), non-profit organizations, health care providers, policymakers, businesses, citizens, and many others working together to protect and improve the health of Iowans. Public health strives to improve the quality of life for all Iowans by assuring access to quality, population-based health services.

One of the most successful population-based health programs is newborn screening.

The Iowa Department of Public Health provides newborn screening for all Iowa newborns through the Iowa Newborn Screening Program (INSP).

In order to provide newborn screening services that best meet the needs and expectations of Iowans, we are asking for your feedback, opinions, and recommendations. Your participation in this community engagement event will provide guidance to INSP staff as they consider new conditions to include on the list of conditions on the screening panel.

On behalf of the Iowa Department of Public Health and the Iowa Newborn Screening Program, I want to thank you for your time and contributions to this important project.

Sincerely,

Gerd W. Clabaugh
Director
Iowa Department of Public Health

What are we talking about?

Introduction

Why should I read this?

This booklet will give you the information you need so you can understand this event and **the Iowa Newborn Screening Program**. It's important that you read this booklet because it will help you prepare for the event, and it will make the event successful.

At this point you're probably thinking all kinds of things… don't worry… you'll have all the information you need when you need it. You may also be thinking "Why did they choose me?" We want to hear from all types of people who live in our state. It's important to us that you have a chance to impact a public health program that affects every family in Iowa.

The information in this booklet was written to explore current research, media, and opinions of professionals and experts. While we tried to discuss many of the issues, it's not possible to explore all the opinions in a short book like this one.

Use this booklet to start thinking about the issues we will discuss at the event, and to develop your own opinions. You may find it helpful to take notes, write questions, and highlight important information as you read.

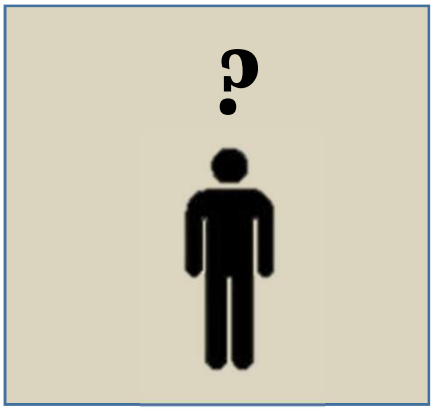
A note about language:
Many of the topics covered in this book are about science and **genetics**. Some of the words you may be reading or hearing for the first time. Check out the dictionary at the back of this book for any new words. Many of these words are also highlighted **blue**.

Chapter 1

Tell me about this event...

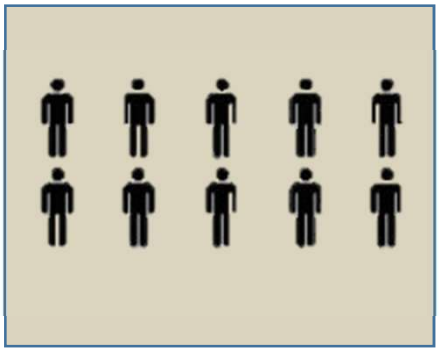
The Iowa Department of Public Health believes that the advice of Iowans like you will help develop policies that serve the public's needs and earn public's trust.

What will I have to do at the event?



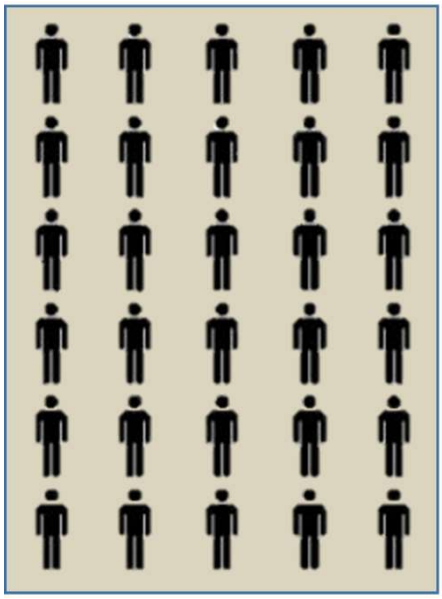
You are here to learn about **newborn screening** and to offer your thoughts and opinions to the Iowa Department of Public Health. Your feedback is extremely important to the future of the program. Please read the information in this booklet carefully over the next month.

What happens at the event?



You will listen to experts, parents, and others who have experience with the Iowa Newborn Screening Program. Trained moderators will help guide discussions with your fellow citizens about important issues. You will discuss these issues both as a large group and in small groups.

Who else will be there?



You are part of a group of about 30 people chosen to represent the population of Iowa. You will meet with your fellow Iowans to discuss the different issues affecting the Iowa Newborn Screening Program. You will be asked to make recommendations based on your own discussions and opinions at the end of the two days.

There will be people available from the newborn screening program to answer questions, and other people who will be there to coordinate the event, take notes and help lead the discussions.

I've never heard of this before. What's the point?

Most people are interested in programs and policies when it affects their health and the health of their community. Unfortunately, there are few opportunities for people to give input on health programs and policies.

A **community engagement event** is a fancy way to say "community discussion." It is based on the idea that the public should have a voice in decisions about public programs. This community engagement event acknowledges that all citizens have important things to say about health policy in Iowa.

Our goal is to get together a group of Iowans who can:

1) learn more about how the newborn screening program makes decisions, and

2) offer advice to the Iowa Department of Public Health.

This booklet is the first step in providing information about the programs. It is important that you read the booklet carefully.

You do not need to be an expert to participate in this event. None of your fellow participants will be experts. During the event, Iowans will meet to discuss their values and opinions. Participants in the event were selected to represent the diverse experiences and perspectives of the people of Iowa.

What is Community Engagement?*

This is an event where people learn about issues, share opinions, and influence a public law or policy. Events like this have helped to inform many different scientific policy issues such as access to healthcare.

The goal is to create respectful and non-threatening discussions where people can learn information, develop and share their values and opinions, and think about others' values and opinions. We heard from the experts and the advocates. We are having this event to hear from you about what Iowans think about these topics.

*For more information on Community Engagement see www.health.state.mn.us/communityeng/

Chapter 2

What are the questions you want me to answer?

The Iowa Department of Public Health (IDPH) is addressing several important matters about the future of the Iowa Newborn Screening Program. This event will discuss four main questions, which are written below.

1. **What are important things to think about when planning for future additions or changes to Iowa's Newborn Screening Panel?**

The Iowa Newborn Screening Program screens for disorders that need to be detected very early. The need for early detection is to provide life-saving or life-prolonging treatments as soon as possible.

The list of the conditions that Iowa screens for is called the Iowa Newborn Screening Panel. New conditions are constantly being considered to be added to the state's newborn screening panel. The question of which conditions to add to the newborn screening panel becomes more complicated as new technologies could allow Iowa to screen for more and more conditions.

2. **How should IDPH make decisions about reporting on new conditions to be added to the Iowa Newborn Screening Panel ?**

As technology and medicine advance, more inherited conditions can be detected through medical screening. There is a concern that several of the new conditions recommended do not meet the criteria for adding a condition to the Iowa Newborn Screening Panel.

Just because conditions can be detected at birth, doesn't necessarily mean that the whole newborn population should be required to be screened for the condition(s). If there is no access to follow-up care or specialists, treatment is not available, or the baby might not develop symptoms until sometime in adulthood, mandatory newborn screening for the condition may not be the best path.

3. How should the IDPH communicate additions to the Iowa Newborn Screening Panel with families?

The IDPH is required to inform families about the newborn screening program, and wants to provide the information in a manner that works best for Iowans. The three conditions that IDPH is considering adding to the newborn screening panel are slightly different than most of the conditions included on the panel.

An infant that screens positive for one of the conditions may not develop symptoms until much later in life. We are still learning about many of these conditions, and some children will be identified as having the condition that never develop symptoms. Also, newborn screening results can be complicated and difficult to understand.

4. How could the community be involved (if at all) in providing ongoing guidance for the Iowa Newborn Screening Program?

The IDPH wants to find ways to increase community involvement in how the newborn screening program operates. This event is one way to gather community input, but it is only a one time event. Are Iowans interested in helping to guide IDPH newborn screening programs in an ongoing way?

Chapter 3

What is newborn screening?

Newborn Screening is a Public Health Program

Newborn screening (NBS) is one of the largest **public health** programs in the country. Newborn screening is offered as a public health service because the benefits of screening are extremely important to the health of babies, families, and society. Nearly 4 million newborns are screened for rare health conditions every year in the United States. Of these, about 12,500 babies (less than 0.1%) are found to have one of these **rare conditions**.

What is public health?
Public Health is the way our government protects and promotes health to the population. Other examples of public health include having safe water, preventing disease, and giving vaccines. Public health programs serve everyone from the population so that everyone benefits from a healthy environment. Many public health programs are required by law.

There are three points of medical (newborn) screening to be aware of:

- It is a process of identifying people within a larger population who have a higher risk for a specific disorder. These people would receive further interventions, like diagnostic testing.
- It is offered to a population that appears healthy
- The primary purpose of the activity is to benefit the individual being screened.

The newborn screen requires a small **blood sample** from the baby's heel. The conditions screened for can have permanent effects on an infant's health. Screening at birth allows start treatment to start as soon as possible, if the baby has been identified as being at risk for having the condition. Treatment could include diet changes, vitamins or enzyme replacement, special medicines, surgery, or specialized medical procedure such as stem cell transplant.

The blood sample is sent to the State Hygienic Laboratory in Ankeny, Iowa for testing. Results of the screening are given to the baby's healthcare provider and the hospital where the baby was born. If there is an abnormal result, **short-term follow-up** nurses contact the baby's healthcare provider to recommend next steps.

Can a parent refuse the Newborn Screening?

In Iowa, if a parent feels the screening goes against their beliefs they may choose to "**opt out**" of newborn screening for their child. They will sign a refusal form provided by their medical provider, which is then sent to the Iowa Newborn Screening Program. In Iowa, about 40 families a year refuse newborn screening for their baby.

What is the federal committee that makes recommendations for newborn screening"?

This federal committee is called the **Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC).** It reports to the US Secretary for Health and Human Services. This committee reviews information about the medical evidence (usual course of the disease over the lifespan, effective treatments available, screening test method available), economic cost-benefits, public health impact, and availability of diagnostic testing, about each new condition. After this evidence-based review, the committee makes recommendations to the Secretary about whether to add the condition to the Recommended Uniform Screening Panel (RUSP). The ACDNC considers the criteria below when making a recommendation.

Based on ACHDNC and the Secretary's recommendation, the RUSP now includes 31 **core conditions** recommend for each state screening panel. They also advise that states consider screening for 26 **secondary conditions.**

More information about the complete list of conditions that are screened in the newborn screening is available in the back of the book (appendix section, page 39).

How does the ACHDNC decide which conditions to add to Newborn Screening?

- The condition should be an important health problem
- The natural history of the condition should be understood
- There should be recognizable early symptoms of the condition
- There should be a test that is easy to preform and interpret, acceptable, accurate, reliable, sensitive, and specific.
- There should be an accepted treatment recognized for the condition.
- Treatment should be more effective if started early
- There should be a policy on who should be treated
- Diagnosis and treatment should be cost-effective
- Finding affected people should be a continual process.

More information on ACHDNC can be found at www.hrsa.gov/advisory-committees/heritable-disorders/index.html

Where do these conditions come from?

In every family there are similarities. You might notice that a cousin has the same smile as their mother, or that you have the same eye color as your grandmother. Newborn screening conditions are no different.

Sometimes the condition is passed through the family by a person who is a **carrier** of the disorder. We call this person a carrier because they are not affected with the condition, but could still pass it on to their children. Children might also have a condition for the first time in the family history, and no one knows why.

A child can **inherit** the condition if:

- They have one or both parents as a carrier. The child may be a carrier of the condition or have symptoms of the condition.
- Something happened unexpectedly during their development while in the womb, making them the first in their family to have the condition.

Either one of these situations can cause a baby to be born with one of the conditions you will learn about through this process.

This information was summarized from "Understanding Genetics" a booklet from the Genetic Alliance and the New York Mid-Atlantic Consortium for Genetic and Newborn Screening Services. ***The entire booklet is linked on our website, and available free at www.geneticalliance.org/publications for anyone wanting more information about genetics.***

Is the newborn screening a diagnosis?

The answer is, "No."

There is a difference between screening and diagnosis. Screening programs find people with health conditions before they show symptoms, and to figure out if they are at risk for getting the condition. Public health programs use screening on the whole population at a specific age or time. The assumption is that most of the people are healthy and the screening results will be normal. If a screening comes back with something abnormal, the person then comes back for more tests. The doctor will look at the tests and health history to determine a **diagnosis.**

Diagnosis

A **diagnosis** requires a test or medical procedure that can confirm a health condition. Identification of a condition can help the healthcare provider know how best to help the person with the condition.

The Iowa Newborn Screening Program screens all babies in Iowa to see if they may be at risk of having one of the conditions on the Iowa Newborn Screening panel. The Iowa Newborn Screening Program does not screen older children, and usually screening program services end after the baby receives a diagnosis or normal result.

How does newborn screening work?

A health provider discusses the screening process with the baby's parents.

A baby has a **heel stick**, which is a poke to give blood, 24-48 hours after birth. The blood is collected on **filter paper**. This may also be called a **dried blood spot test**.

The filter paper is sent to the state laboratory. Laboratory tests are performed to see if the baby has increased risk of nearly 50 conditions.

Negative Screening Result:
No increased risk of these conditions was found. No further follow-up is required. Parents can ask for the results from the child's healthcare provider.

Positive Screening Result:
Pediatricians and parents are notified as soon as possible, usually within a day or two. A **diagnostic or clinical test** may be required.

Diagnostic or clinical test
A test or medical procedure that can confirm or rule out a medical condition in a screening result that is not normal.

False Positive
When a child has an initial/ first test that was not normal, and additional shows a normal result. The child does not have the condition.

False Positive:
The follow-up test is normal. The baby does not have the condition detected by newborn screening. (See page 21 for more details)

True Positive:
The diagnostic test confirms one of the conditions. The baby will be referred to a **specialist**.

Chapter 4
What do we do in Iowa?

In the early 1960's the process for the first newborn screening was invented. This screening was for a condition called PKU. The process that was invented made it cost effective to screen every baby.*

The Iowa legislature passed a law to start the Iowa Newborn Screening Program in 1965. A year later, Iowa started screening babies. Newborn screening is an Iowa Department of Public Health program, and is authorized by Iowa laws (Iowa Administrative Code 641, Chapter 4 and Iowa Code 136A).

Today, there are many people involved in making decisions for what conditions are included in the newborn screening panel, and the process has changed over time. Suggestions to screen for new conditions come from the Iowa Newborn Screening Program, families who have children affected by a condition, advocacy groups, legislators, and/or a federal committee that makes recommendations (see page 11).

The Brief History of Newborn Screening in Iowa**

1965- State law establishes the first newborn screening test. The test was for a condition called PKU.

1966- University of Iowa Laboratory (now called the State Hygienic Laboratory) starts the required newborn screening. Screening is voluntary in Iowa.

1983- Iowa was screening 53% of infants. Screening becomes mandatory (required) for all newborns in Iowa, so all babies are screened.

2003- Iowa starts using a new technology for screening called, "**Tandem Mass Spectrometry**". This technology allows for screening multiple conditions at once. It reduced the cost of screening and could identify 29 new conditions. The number of conditions screened became influenced by cost and technology.

2015- Iowa Department of Public Health Congenital and Inherited Disorders Advisory Committee (CIDAC) brings together a group of experts to determine whether to add Pompe disease (see page 28) to the Iowa Newborn Screening Panel. At the time, CIDAC determined there was not enough evidence to begin screening in Iowa and they advised waiting and asking the public.

2018- Iowa holds a Deliberative Public Engagement Event to develop recommendations for adding **Pompe disease**, **MPS-I** and **X-ALD** to the state screening panel (this is the event you are being asked to participate in).

How much does the Iowa newborn screening cost?

In Iowa, the current cost of the newborn screen is $122. Hospitals and healthcare providers send payment for the screening test to the State Hygienic Laboratory. The hospitals or healthcare providers bill the family (and insurance) for the fee amount, usually included as part of overall newborn care services the family gets billed. The fee is ultimately paid by the newborn's parents using whatever means paid for the birth (for example: paid out of pocket, health insurance, Medicaid, etc.).

The newborn screening fee includes state program administration costs, the cost of the laboratory testing process, transportation of the filter paper card to the laboratory, and the special follow-up services necessary for abnormal results. The newborn screening fee does not cover the cost of any treatment.

A Summary of Newborn Screening in Iowa

- Approximately 39,000 babies are born every year in Iowa.
- Screening of blood samples is done 365 days a year at the State Hygienic Lab of the University of Iowa newborn screening laboratory in Ankeny, Iowa.
- Iowa screens newborns for over 50 inherited conditions, including hearing loss and critical congenital heart disease, in addition to the screening done on blood spots. ***
- Every abnormal newborn screening result is followed by Short-term Follow-up program staff and medical specialists at the University of Iowa.
- Each year in Iowa about 1,850 babies are born with an inherited disorder.

More information...

*More information on the history of Newborn Screening can be found here: http://bit.ly/2b9reje

**See page 38 for a detailed timeline of the Iowa Newborn Screening Program.

***Read the table at the end of this booklet (pg. 39-41) for a detailed description of the disorders screened in Iowa.

Chapter 5

Benefits of the Current Iowa Newborn Screening Program

Detects rare inherited conditions
Most babies who have the rare conditions screened for look healthy and act normally at birth. But these babies could have serious health problems, significant physical or intellectual disability, go into a coma, or even die if their disorder is not detected and treated early.

Prevents some disability and early death
Newborn screening finds **treatable conditions** early so that treatment can begin as soon as possible. Over the past 50 years, newborn screening programs have prevented many babies from developing severe lifelong mental and physical disabilities.

Can prevent diagnostic odyssey
Consider a baby who has a condition, but is not screened at birth. The baby may go on to develop health problems. The parents and doctors may not know what is wrong with the infant. Doctors may have a hard time diagnosing the condition. In certain cases, the baby may require many tests before the doctor and family are able to determine the cause. This is known as a **diagnostic odyssey**. This often comes at great financial and emotional cost. One benefit of screening is preventing a diagnostic odyssey.

Newborn screening is for everyone
Newborn screening is for every baby, no matter how much money your family has. The technology used in newborn screening is sophisticated and time sensitive. The screening offers an incredible amount of health information at an important time in the baby's life, without a family needing to worry about an expensive screening.

Diagnostic odyssey
This describes the long search to find a diagnosis for a patient. A newborn may need many tests to figure out a correct diagnosis. This could include many trips to the hospital or unnecessary treatments.

May detect conditions in siblings or family members

Because inherited conditions are passed down within families (see page 13), screening infants for new conditions can indicate that a sibling or close family member may be affected. The relative may not be having symptoms, but could benefit from further testing.

Improves treatment and access

As conditions are added to newborn screening panels and more and more babies are screened, researchers and doctors learn more about the condition and the natural progression of the symptoms. With this information, better treatment can be developed. After many years, the cost of treatment sometimes decreases and can be easier to access.

Saves costs for families and society

The newborn screening is paid for by families and Iowa citizens through health plans, taxes, and most private insurance. However, the cost of medical treatments and lifelong care for an individual who does not receive early treatment can be expensive. Even though the treatment may be expensive, the cost of living with an untreated condition could be more expensive. Identifying and treating these newborns early can save on future costs from more severe symptoms. Knowledge from newborn screening can help parents make educated decisions about care and their child's future.

Emotional and social benefit

Families who care for children with special health care needs and disabilities will often speak of how much they value their child's smile, their ability to verbally communicate, or their ability to care for themselves. Achieving these "simple skills" may be extremely difficult for these families. Many children who receive early treatment because of newborn screening have a chance at typical child and adult development. These families realize that because of newborn screening, they can interact with their child without thinking about the child's condition. They have a much different relationship with their child.

Chapter 6
Real Stories

Success of Newborn Screening

When **Zach** was born he appeared to be a perfectly healthy baby boy. A week later, Zach was diagnosed with **medium chain acyl-CoA dehydrogenase (MCAD) deficiency** through the Iowa Newborn Screening Program. Infants with MCAD deficiency can die if they develop an infection or go too long without food. Zach is on a high-carbohydrate, low-fat diet, and takes medication to help his body remove toxins and stay healthy. Because of newborn screening and early treatment, Zach now lives a typical teenage life.

Source: http://bit.ly/2eYAwAR

Three days after **Evan** was born he became very sick. His parents did not know what was wrong. Luckily, they received a call from their doctor the same afternoon. Evan's newborn screening results showed that he had **Galactosemia (GALT).** GALT is a condition where the body is unable to properly digest galactose, a sugar found in milk. These sugars build up because they are not able to be used for energy. This can cause speech impairment, eye problems, liver problems, intellectual disability, muscle problems, seizures, or could lead to early death. Understanding GALT and removing dairy from Evan's diet has given him the chance at an ordinary life.

Source: http://bit.ly/2f859rC

Brody and Brooke were both born with **biotinidase deficiency (BD).** People with this condition are unable to recycle the vitamin biotin. This can lead to numerous life-threatening conditions such as seizures, trouble breathing, and the body's ability to fight off common illness. Brody and Brooke were both identified through the Iowa Newborn Screening Program and started taking daily doses of biotin. Their early treatment allows them to live healthy lives.

Source: http://bit.ly/2eYAwAR

Without Newborn Screening

Source: http://bit.ly/2x2FeG5

Giselle was born with **long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD).** Individuals with LCHAD deficiency cannot use stored fats for energy. The fats they eat build up in the heart cells. This can cause serious heart problems, breathing difficulties, coma, and sudden death. With early treatment, individuals can live with reduced health problems on a modified diet. Giselle did not live in a state that did newborn screening for LCHAD and she passed away a few months after birth.

When **Brandon** was born, the disorder **Severe Combined Immunodeficiency (SCID)** was not a part of the newborn screening panel. He was a happy playful baby until he was 6 months old. Like many other babies, he caught a cold. No one knew he had SCID, which prevents the body from fighting off illness. He became tired and weak, was not eating, and continued to get worse. Doctors couldn't identify what was wrong until it was too late. His parents removed him from life support a few weeks later. SCID is one of the most recent conditions added to the Iowa Newborn Screening Panel. Babies identified with SCID typically receive a bone marrow transplant shortly after birth.

Source: http://bit.ly/2h6Ypun

Source: http://bit.ly/1ekZQLS, http://bit.ly/2vWBapr

When **Stephen** was born, there was no newborn screening for **Isovaleric Acidemia (IVA).** IVA is a disorder where the body cannot process proteins, and the build up becomes toxic within the blood and organs. If this continues, it can cause a sudden health crisis involving changes in the blood, seizures, brain swelling, and coma. This happened to Stephen. He lived a healthy and normal life until his health crisis at three years old. Now he lives with neurological damage and complex medical conditions. Stephen's younger sister Caroline also was born with IVA but it was detected through the newborn screening program. She received early treatment and dietary changes which allows her to live a typical life without the health difficulties of IVA.

False Positive Screening

After seven years of infertility, Ann finally had a son, **Gianni**. It seemed that everything was perfect, until she found out two weeks later that he screened positive for **cystic fibrosis**. It would take nearly four months to find out for sure. She was in constant worry about his heath, always living in uncertainty. Finally, Gianni was old enough for the diagnostic test, which came back negative.

Jennifer, another mother, was told that her 10 day old daughter **Sophie**, may have **propionic academia** which required her to have a strict feeding schedule. For two weeks she lived in constant fear and pressure to wake and feed at all hours of the day. Then she found out her daughter was perfectly healthy. Jennifer felt that part of what she was supposed to enjoy with her newborn was taken away from her.

Source: http://on.today.com/2zlJTXU

Late Onset Type

As a child, **Tiffany** developed slowly and was often sick. She seemed clumsy for her age and missed a lot of school. Later, Tiffany developed back problems and doctors did not know what was wrong. At age 11, she was diagnosed with respiratory problems and myopathy, a disease of the muscles. She was told she would probably only live into her 20's. She was constantly visiting doctors and trying new treatments. From ages 12 to 16, Tiffany's health continued to decline, she started using a wheel chair because of back problems, and she eventually had to be home schooled. At the same time, doctors suggested a diagnosis of **Pompe disease** and Tiffany started a clinical trial in the Netherlands for Enzyme Replacement Therapy. Tiffany's health began to improve, but she struggled with changing dosage amounts and the side effects. The therapy wasn't always reliable and she never knew if her condition would improve or get worse. Tiffany says, "The hardest aspect of the disease has been missing out on so much of my adolescence… I get really depressed… I missed out on my 8th grade graduation, lots of dances, dating, learning to drive…It is also really hard to accept it when things that I used to be able to do, I can't do."

Source: http://bit.ly/1Egme7Y

Chapter 7
Challenges Facing Newborn Screening Programs

Some test results will be "false positives"

The newborn screening test is meant to find all babies who might have a condition. To be sure to find everyone, screening tests might call some babies "positive" who are actually healthy. This result is called a "false positive." False positive results happen for many reasons. Different factors change the level of these substances in the baby's blood and affect results. For example, if the blood sample is taken too early after the baby is born, the result will appear abnormal. Babies born prematurely often have false positive results. The number of false positives found through newborn screening depends on the condition. The new conditions may have a higher number of false positives. Every effort is made to reduce the number of false positive results, but they will never be completely eliminated.

Emotional effects of false positives

A newborn with a false positive result is discovered when a diagnostic test is done. However, it may take time to determine that an infant does not have the condition. False positive results may cause anxiety and worry for parents.

This anxiety is often temporary and parents are relieved once diagnostic testing rules out the condition. Other parents may still see their child as having something wrong, whether or not diagnostic testing confirms that their baby is healthy. Parents may treat their child differently or worry unnecessarily.

False positives increase program and family costs

Additional diagnostic testing is always necessary after a positive NBS test. The testing and follow-up care may take many days or months. The costs tend to be small but may be greater if it takes a long time to rule-out a disorder. Families, the state, and the healthcare system often share the costs of follow-up tests. Some families may not be able to afford all of these costs.

If the testing for new conditions increases the chance for false positive or confusing results, people may not believe in the program. If conditions added don't have clear treatment plans, services are spread too thin. Other conditions can't maintain high levels of support services because funding is not always increased with the changes.

Treatment may not be accessible to families

The medical technology to treat new conditions is not available in every state. This is why each state determines which conditions are on the newborn screening. Some treatments are so specific and specialized that families have to travel frequently to services in a different state.

Using insurance to pay for specialized treatment can be complicated, and it can be even more complicated to manage insurance across state boundaries. Some treatments are extremely expensive and not every insurance will cover it, and not every family will be able to afford it. Some insurance may consider the condition as a "**pre-existing condition**."

Screening for Late Onset Conditions creates uncertainty for patients and physicians

New genetic technology can also cause a lot of uncertainty. Many times, genetic results include unclear answers. Sometimes you can't determine whether or not a person has a condition, because there is so much scientists still do not know about genetics.

When the testing does not provide clear results, the patient and doctor have a **diagnostic dilemma.** The doctor may not know how to treat the disorder, or if the patient could benefit from treatment, or what the future would be for their health condition. This can mean a lot of appointments for the patient and the doctor, without any answers.

Diagnostic Dilemma
This occurs when the testing to confirm a condition doesn't have clear results. The doctor may not be able to tell the person they are "normal" but they also may not be able to say "you have the condition."

Parents of children "waiting for a diagnosis" are left with the uncertainty about how to help their child. Every time their child is sick, they wonder if this is when their life will change. Every time their child wants to do anything, they wonder if it will make their condition worse. This can have emotional, financial, social, and opportunity costs for families. http://on.today.com/2zlJTXU

Screening methods may find conditions that are not yet treatable

States try to screen for conditions that have treatments available. As technology expands our ability to screen, newborn screening will find children with conditions that are not treatable. Some people have ethical concerns about finding out about conditions that can't be treated, while others say that they would prefer to know regardless whether treatments exist. State public health programs are required to meet ethical standards, that may be different from what individuals think is best.

Chapter 8
New conditions to consider

Why should Iowa add more conditions to its newborn screening panel?

Recently, the national ***Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)*** (see page 11) recommended the addition of three conditions to state newborn screening panels.

Originally, states only screened for conditions that required immediate action to prevent serious illness or death. Each state decides which conditions to include in their newborn screening panel. Most states screen for the set of core conditions, but some states choose to screen for more.

These recent new conditions have raised some concerns for newborn screening programs around the country. Many states feel that having the technology to screen for a condition does not mean they should start screening right away. The states feel the ethics, potential effects of the screening on families, and availability of treatment within their state should be considered more closely. Many states are also concerned about the number of conditions the committee has recommended in a relatively short time, and the lack of program support and funding to start screening effectively.

What are the three new conditions to consider?

The three conditions ACHDNC recommends adding are:

1. **Pompe Disease**, pg. 24
2. **Mucopolysaccharidosis Type 1** (MPS-I), pg. 25
3. **X-linked Adrenoleukodystrophy** (X-ALD) pg. 26

Many of the current conditions on the newborn screening require the test results within the first few days of life, or the baby could die without treatment. For many of the current conditions, newborns avoid the most devastating effects the condition through specific parent education, special diets or formula, or special vitamins and enzymes. For a few of the conditions, special medical procedures such as a stem cell transplant or surgeries would be required.

The newly recommended conditions may not be life threatening in the first days of life, and might not effect the child until much later in life. The treatments for these new conditions are quite expensive, and may themselves be life threatening.

Image courtesy of Science Direct.com

What is Pompe Disease?

Who's affected?

Pompe disease affects approximately 1 in 30,000 people in the United States. There are two forms of Pompe disease. About 1/3 of cases affect infants (infantile onset), while the other 2/3 affect children and adults (late onset).

What happens to a person's body?

Pompe disease makes the body not able to break down glycogen, which is a form of sugar from the food that's eaten. The sugar builds up in the body and causes problems in the heart and muscles (as well as other parts of the body).

What happens then?

Eventually the heart becomes very large and the person has difficulty walking or moving; difficulty breathing, and may have to use a ventilator. They could experience early death.

How is Pompe disease treated?

There is no cure for Pompe disease. The treatments can help the symptoms be less severe, but any damage already done to the body cannot be reversed. Treatment for Pompe disease would be life-long enzyme replacement therapy. This therapy would be injected into a person's veins every two weeks. The person would have to travel to the specialty clinic for the treatments. Current cost of treatment could exceed $300,000 annually.

Why add Pompe Disease:

- Currently, it is difficult and time-consuming to diagnose Pompe disease, as it can look like other health problems.
- The damage from Pompe disease is permanent. If the baby starts treatment right away, they may live longer, and might have less problems.
- Without treatment, babies with early-onset Pompe disease may die before age 2.
- Pompe disease can be detected through newborn screening.

Why people aren't sure about it:

- Treatments for Pompe disease are more costly than many current conditions and not available throughout the state.
- Pompe disease has a late onset type that affects 2/3 of cases. Newborn screening and further testing may detect a difference between early and late onset, but is not able to determine the severity or time of onset of symptoms for late onset types.
- Late-onset medical tests are considered unethical for other diseases.

Information summarized from www.pompe.org and www.rarediseases.org

What is Mucopolysaccharidosis, Type 1?

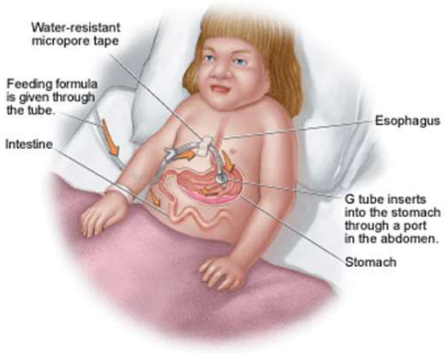


Image courtesy of the National MPS Society

Who's affected?

Mucopolysaccharidosis Type 1 affects about 1 in 100,000 people. It may also be known as MPS-I or **Hurler's Disease.** MPS-I is part of a group of MPS disorders. Each disorder has slightly different symptoms.

What happens to a person's body?

MPS-I can be severe or mild (also called attenuated, pronounced ah-ten-u-ate-ed). MPS-I is similar to Pompe disease in that the body is not able to break down certain sugars, and they build up in the body. The effects are different. MPS-I causes changes in the head and face, hearing loss, and organ problems.

What happens then?

Children with the most severe forms of MPS-I rarely live more than 10 years. Other forms of MPS-I may live through teenage years, adult years, or reasonably "normal" life span, based on the form. The diseases can cause eye problems, extremely short height, stiff joints, problems with speech and hearing, hernias, runny nose, and heart problems.

How is MPS-I treated?

Treatment for MPS-I is a bone marrow (stem cell) transplant and lifelong enzyme replacement therapies.

Why to add MPS-I:

- The damage from MPS-I is permanent. If the baby starts treatment right away, they may live longer, and might have less problems.
- Without treatment, babies with the most severe forms of MPS-I may die before age 10.
- MPS-I can be detected through newborn screening with new equipment.

Why people aren't sure about it:

- Screening detects forms of the disease that will not have symptoms: unknown forms, false positives, and carriers.
- Newborn screening can't tell if a newborn identified with MPS-I will have the severe form of MPS-I or a form without symptoms. – it's difficult to determine true cases.
- Treatments for MPS-I are more costly than many current conditions and not available throughout the state. Families may not be able to travel frequently for treatment.
- There are no protocols developed for patients without symptoms.

Information summarized from www.mps1disease.com

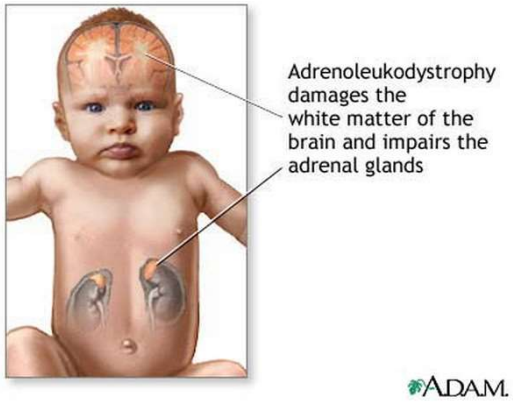


Image courtesy of ADAM.com

What is X-Linked Adrenoleukodystrophy?

Who's affected?

X-ALD affects 1 in 17,000 people. X-ALD affects males differently than females. Females with X-ALD usually develop mild symptoms as they get older. Males develop severe symptoms early in childhood (between ages 4-10).

What happens to a person's body?

People with X-ALD are not able to break down certain fats. These fats build up and make it hard for the nerves to send information to the brain. X-ALD is a **neuromuscular disorder**.

What happens then?

This can affect hearing, vision, muscle strength, swallowing, and leg movement. Some people develop seizures. Later, people can become paralyzed. Eventually it can lead to the body being unable to communicate with the brain and to death.

How is X-ALD treated?

Early treatment through a bone marrow (stem cell) transplant can delay symptoms. However, any symptom of X-ALD cannot be reversed, so if the person does not receive treatment early enough, they may not be able to get it at all. Many of the X-ALD patients who have received bone marrow transplants in clinical trials have died as a result of the transplant itself. There is also some debate about a treatment known as **Lorenzo's oil** (a movie was made about this). Without any treatment, patients die within 2-4 years after developing symptoms.

Why to add X-ALD:

- X-ALD can be detected through newborn screening with new equipment.
- Currently, X-ALD is only diagnosed after symptoms appear. This is often too late for treatment.
- Earlier treatment may save brain function and prolong quality of life.
- The damage from X-ALD is permanent.
- Without treatment, patients typically die within 2-4 years after developing symptoms.

Why people aren't sure about it:

- X-ALD symptoms do not appear in the newborn age (the newborn screening goal).
- A similar condition, Zellweger syndrome, which is fatal and does not have treatment, would be detected with the X-ALD screening.
- Treatments for X-ALD are more costly than many current conditions and not available throughout the state. Families may not be able to travel frequently for treatment.
- Many babies who have received treatment have died from the bone marrow transplant.

Information summarized from www.x-ald.nl

Chapter 9
Three New Conditions to add to the Newborn Screening Panel

Pompe, X-ALD, and MPS1 are different from many of the previous conditions that the ACHDNC has recommended adding to the newborn screening panel.

Some experts have questioned whether screening for these conditions could create more harm than benefit. However, not screening for these conditions could result in children developing disabilities or dying from these conditions. Below is a list of some of the issues to consider when discussing whether or not to add these conditions.

1. **The technology to identify these conditions is available and is relatively affordable to add to newborn screening:** Screening for inherited conditions in newborns is relatively low cost because of the population size and processes available. The same type of screening is unavailable at any other age.
2. **These conditions could be treated more effectively if they were identified earlier**: Developmental affects and disability related to these conditions is permanent, and could cause early death. Early treatment, if effective and available, may delay or prevent the developmental affects and disability of these conditions. It's possible that as more people are detected, treatment will become cheaper and more available.
3. **Treatment accessibility and effectiveness**
   a) **Expensive:** The treatments for these conditions are often experimental, expensive, and difficult to get covered by insurance. These treatments carry more risk, are less accessible, and are more expensive than the treatments for most of the other newborn screening conditions.
   b) **Unclear how effective treatments are:** There is no cure for any of these conditions. Treatment can lessen some of the symptoms, but the person will likely continue experiencing symptoms during their lifetime.
   c) **Difficult to access treatment:** Many of these treatments are specialized and only available in Iowa City. For the transplant, a patient may have to travel to the University of Minnesota.
   d) **Side effects:** Some of the treatments may have significant, life-long side effects for the patients.

4. **Should we screen for conditions that families cannot afford to treat?** The state newborn screening program can only cover the cost of screening (not treatment). There are families currently who struggle to pay for less expensive treatments of currently screened conditions. Many people wonder whether the state should screen for conditions if there is no way to pay for treatments.

5. **Insurability of the children based on results:** Children may also be detected for late onset conditions or conditions that do not have approved treatments. Future insurance coverage (of all types) may be affected by these results. Newborns are unable to make their own choices, and these results may affect their lifelong healthcare options. They may become victims of health and life insurance discrimination.

6. **Ethics of public health:** The role of public health is to reduce mortality and increase overall population health, and not expand medical research. Some experts question whether adding these conditions actually represents research rather than medical care, since so much is still unknown about these conditions.

7. **Newborn screening vs. adult screening:** Each of these three conditions has a late-onset type and milder forms of the condition. Is it the role of newborn screening to identify children who do not develop disease until childhood or their adult years? This could lead to additional testing or medical treatments before they are necessary. This can also lead to "labeling" the child or the adult as having the condition, affecting how they are perceived, or how they may perceive themselves.

Your input on these decisions matters!

In 2015, Congenital and Inherited Disorders Advisory Committee met to look at evidence for Pompe disease screening. They talked about the cost of screening, availability of treatment, and the impact of screening on the community.

The group met for many months, and asked a lot of people for their opinions. After the group voted, CIDAC members and Iowa Newborn Screening Program leadership decided they needed to ask Iowans what they would think about Pompe disease, and other similar conditions.

The community engagement event that you have been selected to participate in is the result of that decision!

Chapter 10

Involving the public in decisions

[WHAT TO DO ABOUT CURRENT AND FUTURE DECISIONS]

The Iowa Department of Public Health must make decisions about whether and how best to add these new conditions to the newborn screening panel as well as future changes. Balancing the benefits of screening with improved health for Iowans is a difficult task.

How does this program involve the public?

Iowa, and a few other states, make decisions for their newborn screening program through consulting with experts, listening to advisory groups, and reacting to new legislative policy. Not all of these states connect with members of the general public community.

In the past, many laws about newborn screening in Iowa have been influenced by advocacy groups. Some of these advocacy groups work with patients and families of children with rare conditions. Other advocacy groups work with pharmaceutical and biotechnology companies that develop treatments for these disorders and would benefit financially.

What are we doing as part of this community engagement event?

Typically, the voices of regular Iowa citizens are not included in these policy recommendations. We are holding this deliberative community engagement to gather your voices. We are inviting you to help us because you can provide a unique perspective and share opinions that represent the values of Iowans.

Your opinions will help the leaders of the Iowa Newborn Screening Program make choices about when and how these new conditions will be screened in Iowa. Your recommendations will also be shared nationally and could change opinions about other conditions in the future.

After the event, we may invite you to consider joining a public advisory group. This group would be consulted in future decisions related to the future of newborn screening in Iowa.

Please think about the issues you have learned about while reading this book. Do you think it's important to ask the public about these topics? Do you think newborn screening should be something that more people talk about?

Chapter 11
What's next & how to prepare

As a part of this group, you will be asked to share your thoughts on the Iowa Newborn Screening Program and what is important for Iowans. Think again about the questions you read in Chapter 2.

1. **What are important things to think about when planning for future additions or changes to Iowa's Newborn Screening Panel?**
2. **How should IDPH make decisions about reporting on new conditions added to the Iowa Newborn Screening Panel?**
3. **How should the IDPH communicate the proposed additions to the Iowa Newborn Screening Panel with families?**
4. **How could the community be involved (if at all) in providing ongoing oversight or guidance for the Iowa Newborn Screening Program?**

We are asking you to listen to your fellow participants during the event and share your own opinions/thoughts. Consider the information that you read, your own opinions, and the opinions of your fellow participants. During the event, you and the other participants will be asked to share recommendations for the IDPH on each of these issues.

Who's listening?

The Iowa Department of Public Health and program leaders for the Iowa Newborn Screening Program created this event. They will be using your thoughts to make future changes for the Iowa Newborn Screening Program. National newborn screening advisors are listening too.

What happens now?

Now that you've read about the issues, you know a little more about what to expect. There's no more homework before you come to the event. You may receive a few more calls or letters to remind you what to bring, where to go, and what to expect at the event.

If you have any questions, please give us a call at 515-725-2290 or email sarah.vangorp@idph.iowa.gov.

The Appendix

(EXTRA DETAILS ABOUT WHAT YOU JUST READ)

A1 Glossary

***These definitions are generalized to fit their purpose within the Newborn Screening Program. For additional definitions related to genetics see the Genetics Home Reference website: https://ghr.nlm.nih.gov/**

A

abnormal result
A newborn screening result that means more testing is needed to see if the baby has a condition

additional screening
To screen for conditions that are not included on the state's newborn screening panel; also called “supplemental screening”

Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)
A federal group that gives advice about the health of babies and children with conditions. Their goal is to reduce illness and death, review new projects related to conditions, and make decisions about adding new conditions.

advocacy group
A group of people who work together to support a cause

B

birth defect
a physical abnormality that is present at or before birth

blood sample
When blood is drawn from the body in order to be tested for medical purposes

bone marrow transplant
A treatment where stem cells from bone marrow, or cord blood are put into the blood and original cells are destroyed. Also known as “stem cell transplant” or “cord blood transplant.” Bone marrow transplants have significant cost and side effects. They are often considered experimental and highly specialized medical procedures.

C

clinical testing
Testing that is done to confirm if a baby has a condition. Not a part of research.

community engagement event
An event that allows the public to come together to talk about an important issue, think of all the different options and make recommendations

confirmatory test
To confirm or rule out a medical condition in a baby with symptoms or a screening result that is not normal. Also called “diagnostic tests.”

C

core condition
A condition that newborn screening is specifically designed to identify
core panel
The list made up of all the conditions that newborn screening is designed to find

D

diagnostic dilemma
This occurs when the testing to confirm a condition doesn't have clear results.
diagnostic odyssey
This describes the long search to find a diagnosis for a patient. A newborn may need many tests to figure out a correct diagnosis. This could include many trips to the hospital.
diagnostic testing
A test done after a baby has a newborn screen that is not normal. This is the test that confirms if the baby has the condition or not. Also called "diagnostic follow-up test" or "confirmatory test."
disorder
An illness that does not allow the body or mind to work how it should
DNA testing
A genetic test that examines DNA. This test may be used to look at DNA changes that can cause disease.
dried-blood-spot testing
The process of testing the small amount of dried blood on the paper used in newborn screening

E

early onset
A condition that shows symptoms early in life. In newborn screening early onset generally means before the age of 1 year.
enzyme replacement therapy
The missing protein is changed and given to the patient to replace their other enzymes

F

false negative result
A result that means that the newborn screening result came back as normal when condition is really present
false positive result
When a child has a follow-up test because the first test was not normal. The follow-up test shows a normal result. The child does not have the condition.
filter paper
A porous paper that is used to store the baby's blood sample for newborn screening

F

"floppy baby" syndrome
A condition in newborns and infants where there is low muscle tone. This is often a sign of Pompe in babies.

follow-up testing
The other tests and procedures done after the initial newborn screening results appear not normal. Follow-up testing confirms if a baby has a condition or not.

G

genetic disease
A condition that is caused by changes in genes or chromosomes. Also known as a "hereditary disease" or an "inherited disorder."

geneticist
A doctor or scientist who studies how genes work and contribute to health conditions

genetic risk
The chance that a person will develop an illness or condition

H

heel stick
When the baby's heel is pricked to collect a sample of blood for newborn screening

hereditary
A condition that can be passed down from parents to their baby. Also known as "inherited."

human genome
All of the DNA a person possesses

Hurler Syndrome
The most severe case of MPS I. Also known as "Hurler-Scheie Syndrome."

I

informed consent
Learning details about a medical treatment to be able to decide whether or not to have the treatment or test.

inherit
To receive a characteristic from a parents' genes

Iowa Department of Public Health (IDPH)
The Iowa Department of Public Health is a state government agency focused on improving the health of people living in Iowa. The newborn screening program is run by IDPH.

L

late onset
The development of symptoms for a condition later in life, compared to a different form of the same condition. In newborn screening, after age 1.

L

leukodystrophy
A set of conditions that affect growth of the myelin sheath, a material that surrounds and protects some nerves

Lorenzo's Oil
A combination of specific oils which may slow the progression of X-ALD

long-term follow-up
The process of keeping in contact with patients identified with a condition through newborn screening. Follow up staff make sure the baby's medical needs are met over a long period of time.

lysosomal storage disorder (LSD)
A group of many conditions that are caused by errors of metabolism (how the body breaks down food) and result in build-up of substances in the cell.

M

MPS
A family of 7 lysosomal storage conditions each caused by a deficiency in a specific enzyme.

MPS I
Caused by a not enough of a lysosomal enzyme. This is an autosomal recessive condition. Also known as "Hurler Syndrome."

N

negative test result
The baby's blood test did not show any signs of the conditions included on the newborn screening panel.

neurologist
A doctor who diagnoses and treats conditions related to the nervous system

neuromuscular disorder
A conditions related to the nerves and muscles

newborn screening
Newborn screening tests babies for serious, but treatable, conditions. It can include a heel stick, hearing screen, and pulse oximetry.

newborn screening panel
A list of conditions that a baby will be screened for after birth. Each state has its own list.

newborn screening test refusal
A parent can refuse screening if the test does not agree with their religious beliefs or practices. The parent must sign a refusal form.

O

opt-out
A parent's right, in some states, to refuse newborn screening for their child

P

Pompe disease
A condition that affects children and adults and involves muscle weakness, an enlarged heart, and breathing problems.

positive screen
This means that the baby's screening test showed signs that they may be at higher risk of having a genetic condition. Follow-up testing must be done to see if they actually have the condition. Also knows as “positive test result” or “out-of-range result.”

public health
Protects and improves the health of a community. Public health focuses on whole populations, not individual patients. Public health programs aim to prevent disease and improve health.

Pre-existing condition
A term used in health insurance for conditions that the patient has received medical advice or treatment for, before enrolling in health insurance plan. There have been times where insurance did not cover pre-existing conditions.

R

rare health conditions
An uncommon condition that affects the ability of the human body to work normally. Also known as “rare disease.”

Recommended Uniform Screening Panel (RUSP)
A recommended screening panel of 32 core conditions and 26 secondary conditions. States use this panel to make decisions about their state screening programs. Also known as “uniform panel.”

residual dried blood spot
The small amount of dried blood that remains on the filter paper cards after newborn screening has been performed. Also known as “residual sample.”

retesting
When a test needs to be done again to explain, confirm or reject the results of the first test

S

screening test
A simple test done on many people to determine the people who might develop a specific disease. A screening test looks for health conditions before symptoms happen.

secondary condition
A condition that is found while looking for another condition. Also known as “secondary disorder.”

S

short term follow-up

The process of making sure all newborns are screened, the doctor is informed of results, other testing has been completed, and that the baby has received a diagnosis and treatment.

specialist

A healthcare provider who has special knowledge about a condition.

stem cells

A cell whose “daughter” cells may change to other cell types

T

tandem mass spectrometry

Tandem mass spectrometry (MS/MS) is the technology that is used to screen newborns for a large number of conditions on the same machine.

testing outcomes

The results you can receive after participating in a test. In terms of Newborn Screening test outcomes, you can test positive, negative, or inconclusive.

treatable condition

A condition with a known treatment that can improve the quality of life or extend life of an individual

true positive result

A small percentage of babies with abnormal test results do have the condition. When these babies get tested again, the result will be abnormal again. The next step is to get the baby treatment.

U

uniform panel

A recommended screening panel of 32 core conditions and 26 secondary conditions. States use this panel to make decisions about their state screening programs. Also known as “recommended uniform screening panel (RUSP).”

X

X-ALD

An inherited condition that occurs mostly in males. It affects the nervous system.

X-linked inheritance

The gene responsible for the disease is found on the X chromosome. This means males are more likely to have the disease then females.

A2 The History of Newborn Screening in Iowa

1965- State Law recommends the first newborn screening test. The test was for a condition called PKU

1966- University of Iowa Laboratory starts the testing.

1979- Testing Laboratory moves to Des Moines. Testing is still voluntary in Iowa.

1980- Iowa starts testing for three more conditions: Galactosemia, MSUD, and Hypothyroidism.

1981- Newborn Screening available for all Iowa infants

1982- Screening became "fee-for-service" after federal grant ends. Results started being tracked with the Iowa Birth Defects Institute at the Department of Public Health.

1983- Iowa was screening 53% of infants. Screening becomes mandatory (required) for all newborns in Iowa.

1984- Newborn Screening was changed from 72 hours after birth to 48 hours.

1988- Screening for Hemoglobin disorders was added after 9 months of pilot testing.

1991- Screening for Congenital Adrenal Hyperplasia was added after 13 months of pilot testing.

1992- Iowa Newborn Screening started helping the North Dakota Newborn Screening program with screening results.

1995- Newborn Screening was changed from 48 hours to 24 hours after birth.

1996- Laboratory testing advanced for more accurate and rapid turn around time with DELFIA technologies.

1999- Follow-up staff started using an internet database to track results.

1999- Laboratory testing advanced with "Tandem Mass Spectrometer" instrument which allowed for testing multiple conditions on the same machine. This increased the accuracy and speed of test results, while decreasing cost. This technology also added screenings for secondary conditions and found more than 29 new conditions. Some of these conditions did not yet have established treatments.

2000- Screening for MCAD was added after a year of pilot testing.

2002- Screening for Biotinidase was added after 6 months of pilot tests.

2003- Iowa starts using the Tandem Mass Spectrometer for Newborn Screenings. Nebraska and North Dakota contract with Iowa to do Newborn Screening. Iowa now screens for over 50 conditions.

2005- Screening for Cystic Fibrosis was considered. Iowa helps with Louisiana's Newborn Screening Program during Hurricane Katrina, and until 2007.

2006- Iowa develops an independent system to pick up screenings from Iowa Hospitals. This courier service decreases the time for testing and results, allowing newborns with time-sensitive conditions to receive services faster.

2007- Screening for Cystic Fibrosis starts. Iowa begins Newborn Screening for South Dakota.

2010- Screening for TYR type 1 (SUAC) begins after a year of pilot testing.

2013- Screening for SCID was added after a year of pilot testing.

2014- Timeliness in Newborn Screening became a goal of the program. This assured babies at risk for a disorder received the option of earliest intervention.

2015- Iowa Department of Public Health brings together group of experts to determine whether to add Pompe to the Newborn Screening Panel. Experts undecided about evidence and readiness.

2018- Iowa holds a Deliberative Public Engagement Event to evaluate concerns for adding Pompe disease, MPS-I and X-ALD.

A3 What are the conditions we screen for in Iowa?

What's it called?	What happens?	Who's affected?*
Amino Acid Conditions 1. Argininemia (ARG 2. Argininosuccinic Aciduria (ASA) 3. Benign Hyperphenylalaninemia (H-PHE) 4. Biopterin Defect in Cofactor Biosynthesis (BIOPT-BS) 5. Biopterin Defect in Cofactor Regeneration (BIOPT-REG) 6. Citrullinemia, Type 1 (CIT), and Type 2 (CIT II) 7. Classic Phenylketonuria (PKU) 8. Homocystinuria (HCY) 9. Hypermethioninemia (MET) 10. Maple Syrup Urine Disease (MSUD) 11. Tyrosinemia, Type I (TYR I), Type 2 (TYR II), Type 3 (TYR III)	• The person's body cannot break down certain amino acids found in some proteins. This causes ammonia and toxins to build up in the body/blood. The body can't get rid of the toxins. • Most people who follow treatment have no/few complications from the condition. • Without treatment, people are at risk for behavioral problems, intellectual disability, muscle problems, vision problems, liver damage, heart disease, brain damage, coma, and/or early death. • **Treatment options**: diet changes and supplements can minimize effects of the condition.	1. 1 in 300,000 people 2. 1 in 70,000 3. Unknown (maybe 15-75 babies in a million) 4. 1 in a million 5. 1 in a million 6. 1 in 57,000 7. 1 in 10,000-15,000 8. 1 in 200,000-300,000 9. Unknown 10. 1 in 85,000 11. 1 in 100,000, 1 in 250,000, unknown
Fatty Acid Oxidation Conditions 1. Carnitine Acylcarnitine Translocase Defficiency (CACT) 2. Carnitine Palmitoyltransferase I Defficiency (CPT-IA) and Type 2 (CPT-II) 3. Carnitine Uptake Defect (CUD) 4. Glutaric Acidemia, Type II (GA-2) 5. Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD) 6. Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD) 7. Medium-Chain Ketoacyl-CoA Thiolase Deficiency (MCAT) 8. Medium/Short-Chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (M/SCHAD) 9. Trifunctional Protein Deficiency (TFP) 10. Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)	• The body cannot break down certain fats they eat into energy for the body. This causes fat to build up in the body and affects muscle tone, behavior, fever, seizures, and breathing. • People who follow treatment can prevent or control the risks of the condition. • People with this condition are at risk for enlarged, weak harts, enlarged liver, low blood sugar, coma, and early death. Without treatment, babies die before age 1. • **Treatment options:** If detected early, diet changes and supplements can minimize the effects of the condition.	1. Unknown, Less than 30 reports 2. Less than 50 total reports 3. 1 in 100,000 4. Unknown 5. Unknown 6. 1 in 15,000 7. Only 1 report, unknown 8. Unknown 9. Unknown 10. 1 in 30,000
Hemoglobin Conditions 1. Hemoglobinopathies (Var Hb) 2. S. Beta-Thalassemia (Hb S/BTh) 3. S. C Disease (Hb S/C) 4. Sickle Cell Anemia (HB SS)	• These conditions affect the shape of the red blood cells in the body. It can cause dehydration and frequent illness. • Sometimes the condition doesn't need treatment, other times the person is at risk for anemia, organ damage, or early death. • **Treatment options:** pain killers, blood transfusions, or antibiotics.	1. 1 in 20,000 2. 1 in 25,000 3. 1 in 835 African American babies 4. 1 in 375 African American babies

Information summarized from www.BabysFirstTest.org

*The number of people affected often varies based on ethnicity and region; this is the US population occurrence.

What conditions are screened now? (Continued)

What's it called?	What happens?	Who's affected?
Endocrine Conditions 1. Congenital Adrenal Hyperplasia (CAH) 2. Primary Congenital Hypothyroidism (CH)	• Endocrine conditions affect the body's ability to produce enough of certain hormones and chemicals which affects healthy growth. 1. CAH is a condition which affects how a person is able to respond to stress, early puberty, and (more rare) the development of the sexual organs (producing male-like genitals in females). 2. CH can affect overall growth and development including intellectual disability, learning disability, and delays in growth and development. • **Treatment options:** genital surgery and steroid treatments.	1. 1 in 15,000 2. 1 in 3,000-4,000
Organic Acid Conditions 1. 2-Methyl-3-Hydroxybutyric Acidemia (2M3HBA) 2. 2-Methylbutyrylglycinuria (2MBG) 3. 3-Hydroxy-3-Methylglutaric Aciduria (HMG) 4. 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC) 5. 3-Methylglutaconic Aciduria (3MGA) 6. Beta-Ketothiolase Deficiency (BKT) 7. Glutaric Acidemia, Type 1 (GA-1) 8. Holocarboxylase Synthetase Deficiency (MCD) 9. Isobutyrylglycinuria (IBG) 10. Isovaleric Acidemia (IVA) 11. Malonic Acidemia (MAL) 12. Methylmalonic Acidemia (Cobalamin Disorders) Cbl A, B) 13. Methylmalonic Acidemia (Methymalonyl-CoA Mutase Deficiency) (MUT) 14. Methylmalonic Acidemia with Homocystinuria (Cbl C,D,F) 15. Propionic Acidemia (PROP)	• These conditions affect the ability to break down certain proteins and some fatty acids. This also affects the nervous system (spine, brain, and reflex). This causes the organic acid and toxins to build up in the body. • Without treatment, this condition can cause breathing problems, brain damage and intellectual disability seizures, coma, and early death. • **Treatment options:** careful diet planning with special formula or supplements based on specific condition	1. 11 cases total 2. Unknown 3. Fewer than 100 worldwide 4. 1 in 36,000-50,000 5. 1 in 200,000 males 6. <1 in a million 7. 1 in 40,000 8. 1 in 87,000 9. Fewer than 30 total 10. 1 in 230,000 11. Fewer than 30 total 12. 1 in 50,000-100,000 13. 1 in 50,000-100,000 14. 1 in 50,000 -100,000 15. 1 in 35,000-75,000
Biotinidase Deficiency (BIOT)	• The body is not able to process a specific vitamin (biotin). This vitamin is used to process the nutrients from food. • Early identification and treatment allows most children with this disorder to have healthy growth and development. • Without treatment, there is a risk for developmental delay, eye problems, and hearing loss. • **Treatment options**: biotin supplements.	• 1 in 60,000

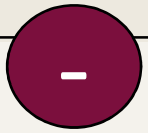
What conditions are screened now? (Continued)

What's it called?	What happens?	Who's affected?
Classic Galactosemia (GALT)	• The body is not able to digest galactose, a sugar in milk. The sugars build up in the blood. • Without treatment, there is a risk for seizures, blood infection, liver damage, or early death. • **Treatment options:** diet changes and taking certain vitamins.	• 1 in 30,000-60,000
Critical Congenital Heart Disease (CCHD)	• There are problems within the development of the heart. This could include problems in the heart's structure or rhythm. • Without treatment babies risk serious disability and early death. • **Treatment options**: special procedures, medicine, or surgery.	• 1 in 4,800
Cystic Fibrosis (CF)	• The body produces extra mucus, which covers all the body organs and tissues. • With treatment, people with CF still live shorter lives, however their symptoms are more manageable. • Without treatment, people with CF experience significant lung damage, digestive problems, growth problems, malnutrition, infertility, and early death. • **Treatment options**: diet changes and taking extra supplements and medications to thin the mucus and properly digest food. People with CF also need special therapy to clear the mucus from the lungs.	• 1 in 3,500 Caucasian • 1 in 7,000 Hispanic • 1 in 17,000 African American
Hearing Loss (HEAR)	• The ear is not able to hear and process the sounds of the environment, to varying degree. • This affects the person's ability to speak, understand language, and express themselves socially. • **Treatment options:** audiology, technology and working with a specialist.	• 3 in 1,000
Severe Combined Immunodeficiency (SCID)	• The body is not able to fight off serious infections. The immune system does not function. • Children die before age 2 without effective treatment. • With transplant children can live healthy lives. • **Treatment options**: isolation, a specific type of medical treatment, or bone marrow transplant.	• 1 in 40,000-75,000

A4 Other conditions that the Advisory Group considered?

What's it called?	Why was it not accepted to newborn screening?
Guanidinoacetate Methyltransferase Deficiency	Screening test is not yet valid; few cases found in newborns.
22q11 Deletion Syndrome	Population data not available; screening test not yet valid; unknown treatment effects.
Hyperbilirubinemia/ Kernicterus	Unclear about benefit to newborn screening vs. clinical screening, unclear cost savings
Hemoglobin H Disease	Evidence needed: late onset types; effect of treatment; infrastructure expectations, population data
Spinal Muscular Atrophy (SMA)	(currently in 2nd review) Additional pilot study data and assessment of therapy needed
Krabbe Disease	Treatment is costly with negative side effects; screening test needs to be refined
Fabry Disease	Common late onset of disease (older than age 10); negative side effects of treatment; unknown effect of newborn treatment
Neiman-Pick Disease	Population data not available; unknown treatment effects; no pilot study data

A5 What do the screening results mean?

Testing outcomes you might hear	What it means
Positive Screening  Next Steps: 1. The baby's doctor and parents are told about the results. The doctor may do other tests to decide on a diagnosis. 2. If the doctor's extra tests show the baby has a disorder, then the doctor gives the baby a diagnosis or name for the medical condition. 3. The baby and family will work with a specialized medical team to learn more.	A possible medical condition was detected
Negative Screening  Next Steps: Parents can ask about the results from their baby's doctor. They do not receive any phone calls about the test. (Note: it is possible, though unlikely, for a baby to have a negative screening and then later find out the condition was missed. The condition would be detected through routine medical care after symptoms develop)	The test did not detect any of the 50 medical conditions
Unclear Results  **(Possibilities: False Positive, Carrier, Late-Onset Type, Indeterminate result, Variant of Unknown Significance)** Next Steps: 1. The baby's doctor and parents are told about the results. The doctor will do other tests and procedures to determine if there is a disorder. 2. The family will work with a specialized medical team to learn more. Possibilities: • False Positive: Follow up for the results showed the baby does not have the condition, or the baby may be a carrier. This is called a false positive. • Late Onset Type: The doctor's extra tests show the baby may have a condition, but the results are unclear. Baby may have symptoms that appear later in life. • Carrier: The extra tests show that the baby may have the condition, but may never have symptoms. There may be a chance the baby passes the condition to future children.	The results are unclear

Notes and questions:

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